

The main health risk of PTEN hamartoma tumor syndrome (PHTS) is cancer, but being born with a PTEN mutation does not mean you will get cancer, or have other health or learning problems associated with this mutation. People can have exactly the same type of PTEN mutation yet be affected very differently. A combination of diet, lifestyle, environment and overall genetic makeup may be the reason for this. Currently, screenings offer the best chance of finding cancer early, and can help to stop some cancers from developing in the first place.

For Women

From age 18

- ▶ breast self-exam: monthly

From age 25

- ▶ breast exam by a doctor: every 6 to 12 months

From age 30-35

- ▶ breast X-ray and, if advised, breast MRI: yearly
- ▶ transvaginal ultrasound of the uterus and cell samples from the womb lining (endometrium) if advised: yearly

Connect * Collaborate * Cure

For All Adults

Yearly

- ▶ physical exam
- ▶ skin check
- ▶ thyroid ultrasound

From age 35

- ▶ colonoscopy: every 5 years or more often if needed

From age 40

- ▶ kidney scan if advised: yearly or every other year

Ask your doctors about:

- * the common warning signs of cancer.
- * surgeries which can help to lower the risk of developing some types of cancer.
- * the best plan for managing your health risks. If you have symptoms and/or a family history of cancer, you may be asked to start screenings earlier and have other screening too.

For Anyone Under 18

Yearly

- ▶ physical exam
- ▶ skin check
- ▶ thyroid ultrasound

If advised:

- ▶ learning and development checks
- ▶ assessment for autism
- ▶ brain MRI

Connect * Collaborate * Cure

PTEN Hamartoma Tumor Syndrome

PHTS is a rare but under-diagnosed genetic syndrome which affects about 1 in 200,000 people. It is caused by a PTEN gene not working properly, and this raises the risk for developing benign growths and certain cancers. For some people, it can affect learning and development.

Our Mission

- * Connect and support PHTS patients worldwide
- * Work with clinicians who have expert knowledge of PHTS to develop guidelines for Clinical Centers of Excellence
- * Work with researchers to find treatments, and offer patients opportunities to be engaged in this process
- * Collaborate with partners who share our vision for supporting patients

Connect * Collaborate * Cure

PTEN Hamartoma Tumor Syndrome Foundation



in collaboration with
PTEN Italia



ptenfoundation.org

ptensyndromefoundation@gmail.com



The content of this leaflet is provided for education and information purposes only. For advice about diagnoses, treatments and health screenings, please consult qualified medical professionals.

The National Comprehensive Cancer Network's screening guidelines for PHTS are regularly reviewed. Please check their website for the latest information about screenings.

PTEN Hamartoma
Tumor
Syndrome
Foundation

PTEN Hamartoma Tumor Syndrome

Managing Health Risks



www.ptenfoundation.org